CLAIMS

What is claimed is:

1. A method for detection of a variant nephroretinin polypeptide in a subject, comprising:

a) providing a biological sample from a subject, wherein said biological sample comprises a nephroretinin polypeptide; and

b) detecting the presence or absence of a variant nephroretinin polypeptide in said biological sample.

2. The method of Claim 1, wherein said variant nephroretinin polypeptide is a C-terminal truncation of SEQ ID NO:2.

3. The method of Claim 2, wherein said variant nephroretinin polypeptide is selected from the group consisting of SEQ ID NOs: 6, 10, 12, 14, 16, and 20.

4. The method of Claim 1, wherein the presence of said variant nephroretinin polypeptide is indicative of nephronophthisis type 4 kidney disease in said subject.

5. The method of Claim 1, wherein said biological sample is selected from the group consisting of a blood sample, a tissue sample, a urine sample, and an amniotic fluid sample.

6. The method of Claim 1, wherein said subject is selected from the group consisting of an embryo, a fetus, a newborn animal, and a young animal.

7. The method of Claim 6, wherein said animal is a human.

8. The method of Claim 1, wherein said detecting comprises differential antibody binding.

9. A kit comprising a reagent for detecting the presence or absence of a variant nephroretinin polypeptide in a biological sample.

10. The kit of Claim 9, further comprising instruction for using said kit for said detecting the presence or absence of a variant nephroretinin polypeptide in a biological sample.

11. The kit of Claim 9, further comprising instructions for diagnosing nephronophthisis in said subject based on the presence or absence of said variant nephroretinin polypeptide.

12. The kit of Claim 9, wherein said reagent is one or more antibodies.

13. The kit of Claim 12, wherein said antibodies comprise a first antibody that specifically binds to the C-terminus of said nephroretinin polypeptide and a second antibody that specifically binds to the N-terminus of said nephroretinin polypeptide.

14. The kit of Claim 9, wherein said variant nephroretinin polypeptide is a C-terminal truncation of SEQ ID NO:2.

15. The kit of Claim 14, wherein said variant nephroretinin polypeptide is selected from the group consisting of SEQ ID NOs: 6, 10, 12, 14, 16, and 20.

16. A method for detection of a variant inversin polypeptide in a subject, comprising:

a) providing a biological sample from a subject, wherein said biological sample comprises an inversin polypeptide; and

b) detecting the presence or absence of a variant inversin polypeptide in said biological sample.

17. The method of Claim 16, wherein said variant inversin polypeptide is a C-terminal truncation of SEQ ID NO:22.

18. The method of Claim 17, wherein said variant inversin polypeptide is selected from the group consisting of SEQ ID NOs: 24, 26, 28, 30, 34, 36, 38 and 40.

19. The method of Claim 16, wherein the presence of said variant inversin polypeptide is indicative of nephronophthisis type 2 kidney disease in said subject.

20. The method of Claim 16, wherein said detecting comprises differential antibody binding.